Olivopontocerebellar Atrophy Presenting with Stridor

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Abstract

The spectrum of degenerative ataxia includes the symptomatic degenerative ataxias and the primary degenerative ataxias. The later may be sporadic and idiopathic or hereditary, being genetically determined. When an individual ataxic patient presents with an adult-onset degenerative ataxia and has a negative family history, the physician is faced with a diagnosis of pure idiopathic sporadic degenerative ataxia or one of the hereditary ataxias.

The clinical spectrum of olivopontocerebellar atrophy (OPCA) usually consists of pancerebellar signs with pyramidal and abnormal eye movements. Although Stridor is more commonly found in multisystem atrophy, it is rarely seen in OPCA. We, here report a case of third decade onset of ataxia presenting with stridor.

INTRODUCTION

In 1900 Dejerine and Thomas first introduced the term olivopontocerebellar atrophy (OPCA). Since then, the classification of idiopathic acquired ataxias1,2 has evolved a great deal. The initial two cases of Dejerine and Thomas involved two middle-aged patients with chronic progressive cerebellar degeneration and autopsy findings of gross atrophy of pons, cerebellum, middle cerebellar peduncle and inferior olives. OPCA has not proven to be a single entity. OPCA overlaps with spinocerebellar atrophy and multisystem atrophy (MSA). Clinical distinction between these entities is based on the dominant clinical feature, which may be cerebellar ataxia (OPCA, MSA and SCA), Parkinsonism (MSA), or autonomic features (MSA).

CASE REPORT

A 45 years right handed male, non-addict was admitted with history of acute stridor. He was diagnosed to have bilateral abductor cord palsy of the vocal cords and hence underwent an emergency tracheostomy and was later referred for neurology opinion.

On enquiry the patient gave history of simultaneous, gradual onset, progressively increasing weakness with decrease in muscle mass of both the lower limbs. He had a history of difficulty in getting up from the squatting position since the past 11 years. He also gave history of difficulty in walking with swaying to the left side, stiffness in both the lower limbs and tripping on walking, gradually progressing since 11 years. There was no history suggestive of fasciculations, weakness in the upper limbs, bladder or bowel incontinence, headache, vomiting, convulsions, trauma to the head or spine.

He had a history of breathlessness and hoarseness of voice since two years, gradually increasing, now presenting with inspiratory stridor. He did not have any symptoms of dysphagia, nasal regurgitation, decreased vision or history suggestive of any other cranial nerve abnormalities.

Family history was not suggestive of similar complaints in 1st or 2nd degree relatives. He was born of a non-consanguineous marriage and both his parents, aged more than 50 years, did not exhibit such symptoms.

On examination, his vital parameters were within normal limits, without any evidence of resting tachycardia or orthostatic hypotension. General examination was also normal. central nervous system examination revealed that his higher functions were normal with a Mini Mental Status Score of 29/30. Examination of the cranial nerves revealed slowed and broken up pursuits and saccades bilaterally, more on the left side, and bilateral fasciculations of the tongue. Indirect laryngoscopy revealed vocal cords in the paramedian position. Motor system examination revealed that all muscle groups showed wasting with normal tone, power being 4/5 in all muscle groups in all four limbs. Superficial reflexes were normal except for jaw jerk which was brisk, plantars were bilateral flexors. Deep tendon reflexes were all exaggerated. Hoffman’s, Trommer’s and Wartenberg’s sign were positive bilaterally. Superficial, and deep as well as cortical sensations were normal. He exhibited postural and intention tremors.
bilateral. Dysdiadokokinesia, finger nose ataxia, rebound phenomenon, and heel-knee test were present bilaterally. Other systemic examination was unremarkable.

On laboratory examination, his Hb was 14.6 gm%, with a WBC count of 5200/mm³, and ESR of 4 mm/hr, his LFT/RFT were within normal limits.

His T₃, T₄, TSH were within normal limits. In view of the clinical features we did a MRI which revealed changes suggestive of olivopontocerebellar degeneration in the brain with atrophy of the pons, cerebellum (Fig. 1) and spinal cord in the mid and lower dorsal region of the spinal cord. EMG findings revealed no evidence of widespread denervation to suggest an anterior horn cell disease. Fibrillations and few polyphasic large amplitude potentials in the tongue suggested denervation, due to the localized pathology in the brain stem.

**DISCUSSION**

We had a 40 years male with third decade onset of pancerebellar signs with pyramidal involvement (brisk reflexes, jaw jerk present) and abnormal eye movements with no extrapyramidal involvement or autonomic features and no family history, presenting with stridor.

Symptomatic causes of ataxia such as drug toxicity (e.g. phenytoin, lithium and lead toxicity), chronic alcoholism, hypothyroidism (normal thyroid function tests), paraneoplastic syndrome (X-ray chest and ultrasonography of abdomen and pelvis normal) and space occupying lesions (SOL) in the brain (MRI- no SOL) were ruled out. The lack of family history precluded a diagnosis of a dominant cerebellar ataxia. Autosomal recessive ataxias such as Friedreich’s ataxia, ataxia telangiectasia, vitamin E deficiency and Refsum’s disease were ruled out on the basis of third decade presentation and lack of other associated features which are typical of each of these ataxias.

When an individual has ataxia without any family history, he could have a pure idiopathic sporadic ataxia or an atypical late onset Friedreich’s ataxia (polyneuropathy and areflexia accompanying ataxia) which was not seen in our patient, where family history may not be present as it is in an autosomal recessive condition. It could be possible that he could even have spinocerebellar ataxia (SCA) types 2 and 6. In our patient SCA-2 was ruled out as there was no ataxia of speech and no hyporeflexia. Similarly SCA-6 was ruled out as the onset of the disease is usually in the fourth to the fifth decade with nystagmus being observed which were lacking in our patient.

Other diagnoses such as multiple system atrophy (MSA) and corticobasal ganglionic degeneration were also considered. Corticobasal ganglionic degeneration was ruled out in view of absence of typical features such as apraxia, bradykinesia and cognitive deterioration. Multiple system atrophy was considered in view of sporadic onset, presentation with stridor, and prominent cerebellar signs. However, no features of autonomic dysfunction or striatonigral degeneration were present, to justify a diagnosis of MSA. However a possible progression to MSA over time would have to be kept in mind in this patient.

The gross appearance of the brain is characteristic in all cases of OPCA. Neuronal degeneration in the arcuate, pontine, inferior olivary nucleus and pontobulbar nuclei as well as in cerebellar cortex is also present. The pons is diminutive in size, especially in area of basis pontis. There is also loss of Purkinje cells in cerebellum with preservation of dentate nucleus. The substantia nigra of the midbrain shows evidence of tissue loss. Anti- Purkinje cell antibodies are the diagnostic markers in OPCA. In sporadic OPCA, oligodendroglial and neuronal intracytoplasmic and intranuclear inclusions are frequently seen, whereas in autosomal dominant OPCA, lesions are found in spinal cord, especially in the posterior columns, spinocerebellar tracts and anterior grey horn cells. The cerebellar features are less prominent. Stridor seen in OPCA is due to paralysis of laryngeal abductors. In patients with stridor, only 7% were found to have an underlying neurological condition and in this only 1-2% of patients had MSA as the etiology. Duration of familial OPCA is 15 years and that of sporadic OPCA is six years. OPCA is frequently associated with small cell carcinoma and ovarian carcinoma. MRI is the imaging of choice revealing pancerebellar and brain stem atrophy, with flattened pons and enlarged fourth ventricle and cerebellopontine angle and demyleination of transverse pontine fibers.

Although the emergency management of stridor is tracheostomy, the choice of treatment for patients with established bilateral abductor cord palsy is varied, but no operation should be attempted till at least two months after the onset of paralysis, thus allowing for any possibility of spontaneous recovery, unless detailed EMG studies reveal complete denervation. A permanent speaking valve tracheostomy may be the best choice in the professional voice user, other interventions which have a limited role, are the many variants of arytenoidectomy, cordectomy and reinervation surgeries.
REFERENCES


Announcement

The Annual Conference of Indian Rheumatology Association, IRACON-2003, is being held at New Delhi from November 27-30, 2003.

Additional details may be obtained from the Organising Secretary, Dr. Rohini Handa, Clinical Immunology and Rheumatology Service, Department of Medicine, All India Institute of Medical Sciences, New Delhi - 110029, India. Tel. : 011-2659 4993; Fax : 011-2658 8663; E_mail : secretariat@iracon2003.com Website : www.iracon2003.com

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